



## HapMap Tutorial Exercises

### ASHG meeting 2007

#### Exercise 1. Searching SNP details

Search for snp: rs2211792. Name the alleles for this SNP in the forward strand relative to the reference genome sequence. What gene does this SNP map to? Is this SNP polymorphic in all four populations? Consider that a minor allele frequency  $> 0.20$  indicate a SNP is common, is this a common or rare SNP?

#### Exercise 2. Finding tag SNPs

Polymorphisms in the CLOCK gene are associated with sleep disorders. To design SNP assays in a German population, find the minimum set of common SNPs that will capture all haplotypes. [Hint: tagSNPs]

#### Exercise 3. SNPs and LD

Search for the ABO gene in rel21a (NCBI B35 data source). Turn on the recombination rate, and all the analysis tracks. How many haplotype blocks can you visually identify? Download the LD for the following SNPs in the CEU population:

rs8176720

rs626035

rs2073827

rs644234

[Hint: Use HapMart rel21a CEU dataset to download LD data for a list of SNPs]

#### Exercise 4. HapMart

In the HapMap-YRI population, find SNPs located in the 3'-UTR of the BRCA2 gene. Report the genotype frequency for the SNPs.

#### Exercise 5: Searching by disease

Search for "sickle cell anemia" on the HapMap browser (B36 data source).

- What chromosome regions does the search result show?
- Click on mutgsys468 on chr13. What gene(s) does this region enclose?
- What pathway is affected by the mutation in this gene? [Hint: Turn on the

Reactome tracks]

#### Exercise 5: Searching by disease (cont)

- Bring up the entire gene on the HapMap browser [Hint: Enter gene name in the search field]
- Download a report with tag SNP details for this region using the default configuration settings. For the list of tag SNPs, download the assay details for CEU population. [Hint: Use HapMart to extract the assay details for a list of SNPs]

#### Exercise 6: Searching annotated GWA hits

Search for all GWA hits on chromosome 1.

[Hint: gwa\* - an asterisk can be used as a wildcard]